

WHAT IS CLAIMED IS:

1. An isolated, purified, or recombinant polynucleotide comprising a contiguous span of 8 to 50 nucleotides of any one of SEQ ID Nos 1, 2, 4 or the complement thereof, wherein said span includes a AA4RP-related biallelic marker in said sequence.

5 2. The isolated, purified, or recombinant polynucleotide of Claim 1, wherein said polynucleotide is selected from the group consisting of:

10 (a) an isolated, purified, or recombinant polynucleotide comprising a contiguous span of at least 12 nucleotides of SEQ ID No 1 or the complements thereof, wherein said contiguous span comprises at least one of the nucleotide positions of SEQ ID No 1 selected from the group consisting of a T at position 1239, a ~~T~~ at position 12347, a C at position 13269, an A at position 13475, a T at position 15241, a G at position 42218, an A at position 45442, and a T at position 77058;

15 (b) an isolated, purified, or recombinant polynucleotide comprising a contiguous span of at least 12 nucleotides of SEQ ID No 1 or the complements thereof, wherein said contiguous span comprises at least 10 consecutive nucleotides of at least one of the nucleotide positions of SEQ ID No 1, wherein said positions are selected from the group consisting of 12947 to 12958, 13470 to 13526, 13641 to 13752, and 14271 to 15968;

20 (c) an isolated, purified, or recombinant polynucleotide comprising a contiguous span of at least 12 nucleotides of SEQ ID No 4 or the complements thereof, wherein said contiguous span comprises at least one of the nucleotide positions of SEQ ID No 4 selected from the group consisting of a T at position 319, a C at position 1241, an A at position 1447, and a T at position 3213;

25 (d) an isolated, purified, or recombinant polynucleotide comprising a contiguous span of at least 12 nucleotides of SEQ ID No 4 or the complements thereof, wherein said contiguous span comprises at least 10 consecutive nucleotides of at least one of the nucleotide positions of SEQ ID No 4, wherein said positions are selected from the group consisting of 919 to 930, 1442 to 1498, 1613 to 1724 and 2243 to 3940;

(e) an isolated, purified, or recombinant polynucleotide comprising a contiguous span of at least 12 nucleotides of SEQ ID No 2 or the complements thereof;

(f) a polynucleotide according to (e), wherein said contiguous span comprises a T at position 1153;

30 (g) a polynucleotide according to (f) wherein said contiguous span comprises at least 10 consecutive nucleotides selected within positions 21-1121;

(h) an isolated, purified, or recombinant polynucleotide wherein said contiguous span is 18 to 35 nucleotides in length and said biallelic marker is within 4 nucleotides of the center of said polynucleotide;

(i) a polynucleotide according to (h), wherein said polynucleotide consists of said contiguous span and said contiguous span is 25 nucleotides in length and said biallelic marker is at the center of said polynucleotide; and

5 (j) an isolated, purified, or recombinant polynucleotide, wherein the 3' end of said contiguous span is located at the 3' end of said polynucleotide and said biallelic marker is present at the 3' end of said polynucleotide.

3. A recombinant vector comprising a polynucleotide of Claim 1.

4. A host cell comprising a recombinant vector according to claim 3.

5. A non-human host animal or mammal comprising a recombinant vector according to claim

10 4.

6. A non-human host animal or mammal comprising an AA4RP gene disrupted by homologous recombination with a knock out vector, wherein said vector comprises a polynucleotide of Claim 1.

7. A method of genotyping comprising determining the identity of a nucleotide at a AA4RP-related biallelic marker or the complement thereof in a biological sample.

15 8. A method according to claim 7, further comprising amplifying a portion of said sequence comprising the biallelic marker prior to said determining step.

9. A method according to claim 7, wherein said determining is performed by an assay selected from the group consisting of a hybridization assay, a sequencing assay, a microsequencing assay, and an enzyme-based mismatch detection assay.

20 10. A method of estimating the frequency of at least one allele of at least one AA4RP-related biallelic marker in at least one population comprising:

a) genotyping individuals from said population for said biallelic marker according to the method of claim 7; and

b) determining the proportional representation of said biallelic marker in said at least one population.

25 11. A method of detecting an association between a genotype and a trait, comprising the steps of:

(a) determining the frequency of at least one AA4RP-related biallelic marker in trait positive population according to the method of claim 10;

30 (b) determining the frequency of at least one AA4RP-related biallelic marker in a control population according to the method of claim 10; and

(c) determining whether a statistically significant association exists between said genotype and said trait.

12. A method of estimating the frequency of a haplotype for a set of biallelic markers in at least one population, comprising:

(a) genotyping at least one AA4RP-related biallelic marker according to claim 7 for each individual in said at least one population;

5 (b) genotyping a second biallelic marker by determining the identity of the nucleotides at said second biallelic marker for both copies of said second biallelic marker present in the genome of each individual in said at least one population; and

(c) applying a haplotype determination method to the identities of the nucleotides determined in steps (a) and (b) to obtain an estimate of said frequency.

13. A method according to claim 12, wherein said haplotype determination method is selected from the group consisting of asymmetric PCR amplification, double PCR amplification of specific alleles, the Clark algorithm, or an expectation-maximization algorithm.

10 14. A method of detecting an association between a haplotype and a trait, comprising the steps of:

15 (a) estimating the frequency of at least one haplotype in a trait positive population according to the method of claim 13;

(b) estimating said frequency of said haplotype in said control population according to the method of claim 13; and

(c) determining whether a statistically significant association exists between said haplotype and said trait.

16. An isolated, purified, or recombinant polynucleotide that encodes a polypeptide comprising a contiguous span of at least 6 amino acids in SEQ ID No 3.

20 17. An isolated, purified, or recombinant polypeptide comprising a contiguous span of at least 6 amino acids of SEQ ID No 3.

18. An isolated or purified antibody composition that selectively binds to an epitope-containing fragment of a polypeptide of claim 16.